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Research Paper

Goldenhar Syndrome

Medical Science

KEYWORDS : Congenital abnormalities. Goldenhar syndrome. Oculoauriculovertebral syndrome, epibulbar lipodermoid.

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ABSTRACT

Goldenhar syndrome is one of the most common congenital anomalies of the first and second branchial arches. Its main alterations affect the eye (dermoids and/or epibulbar lipodermoids), the external ear (auricular appendages, blind-ended fistulas) and the spine (hemivertebrae and vertebral fusion, among other malformations). This work presents the 1 yr 2 month old male child with Goldenhar syndrome. Literature and etiopathogenesis are also briefly reviewed.

Introduction

Goldenhar syndrome (GS) is also known as hemifacial microsomia, oculo-auriculo-vertebral (OAV) syndrome, facio-auriculovertebral syndrome, Goldenhar-Gorlin syndrome, first and second branchial arch syndrome, and oculo-auriculo-vertebral dysplasia. GS was named in 1952 when Goldenhar described a number of facial problems that tend to occur together. Oculoauriculovertebral dysplasia is a congenital symptomatic complex of unknown etiology, in which the primary alterations are located on the eye (dermoid and/or epibulbar lipodermoid), the external ear (auricular appendices, cervical sinus) and the spine (hemivertebrae, vertebral fusions, and other malformations)[1]. Other anomalies have been documented in association with Goldenhar complex, including cardiac, pulmonary and renal defects.[1]

This syndrome comprises of malformations arising from defects in the 1st and 2nd branchial arches. GS is a condition with a prevalence ranging from 1:3500 to 1:7000 live births, and a male-female ratio of 3:2[2] . However, the exact genetic defect is not known.[2] The use of some drugs such as cocaine, thalidomide and retinoic acid during pregnancy, as well as maternal diabetes; have been reported as predisposing factors for the development of oculoauriculovertebral dysplasia.[3]

CASE REPORT

1 yr 2 month old male child presented with cleft lip ,high arched palate, macrostomia and microtia on right side, with preauricular tags on both sides, limbal dermoid with dermal lipoma of right eye . The child had mild motor delay and development was normal in other domains. On systemic examination no abnormality was found. Investigation further revealed ,bilateral mild conductive hearing loss, lateral cervical spine x ray revealed posterior fusion of C2 and C3, x ray of maxilla showed absent condyle on right side ,2-D echo showing ASD ostium secundum of 2 mm size.

Discussion

Goldenhar syndrome (GS) is a well-recognised developmental disorder involving first and second branchial arches and is characterized by considerable phenotypic variability.[1]

Etiology and embryopathogenesis

Majority of cases are sporadic and there is a very small chance of familial occurrence. In many reports autosomal recessive or dominant inheritance has been suggested.[4] Multifactorial inheritance (interaction of many genes, possibly in combination with environmental factors such as chemical exposure). Early developmental arrest in 4th week probably due to decrease in blood flow, drugs such as primidone and vitamin-A and diseases such as diabetes, haemorrhagic incidence during 8th to 12th week involving 1st and 2nd branchial arches, chromosomal non-dysjunction and subsequent mesodermal tethering might be the underlying mechanisms.

Laboratory investigations

Currently no genetic/DNA test is available; hence prenatal diagnosis is not possible. Good quality ultrasound may detect the obvious defect. High resolution computed tomography especially for inner ear, middle ear and vertebral defects and magnetic resonance imaging might be helpful in the diagnosis

Treatment

Children with Goldenhar syndrome often need many surgeries including plastic surgery to fix the jaw, cheeks and ears. Specialized dental care, hearing aids, speech therapy, physiotherapy may be required.

Prognosis

Low IQ, systemic involvement may show guarded prognosis. Otherwise a normal life span is expected.

In summary, the molecular basis of GS is still unclear, and currently no specific diagnostic test is available.*P*atients with GS can have multiple congenital anomalies, and they need particular attention to internal abnormalities. Pediatric specialists should consult with ear-nose-throat, orthopedics, neurosurgery, and ophthalmology clinics to decide on the most appropriate treatment plan, which varies with age and systemic associations. Dental care with experienced multidisciplinary team of orthodontists and maxillofacial surgeons is also necessary for good results[5]. Treatment of patients with mandibular aplasia is complex and can be made with rib grafts, whereas underdeveloped mandibles can be lengthened with bone distraction devices[5]. The management of GS patients requests a long-term commitment, and involves multiple procedures spanning the child's period of growth and development.

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Fig 1. Macrostomia with cleft lip

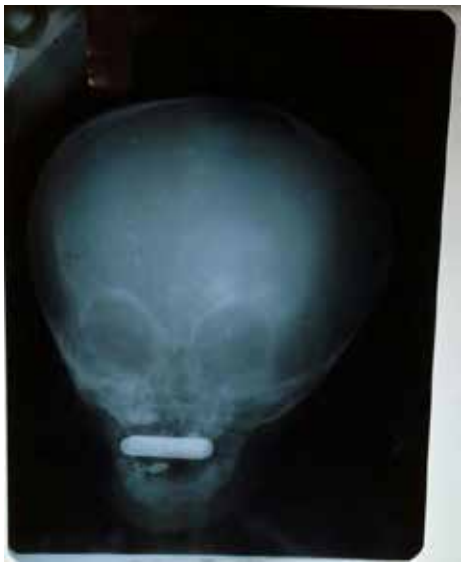
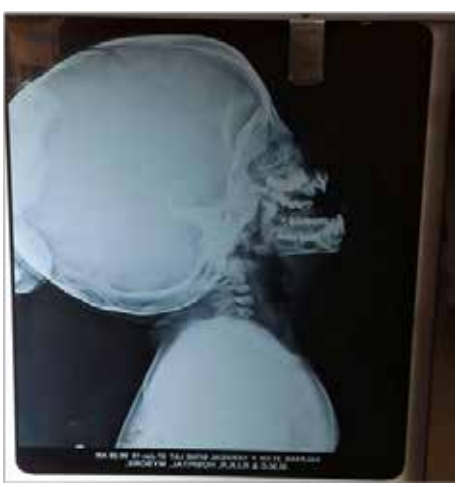


Fig 2. Preauricular tags on left side

Fig 3. X- ray skull showing absent right condyle

Fig 4. Preauricular skin tag & limbal dermoid on rt eye.

Fig 5. X-ray lateral cervical spine showing posterior fusion of C2 –C3.



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